

16	503.5	28.1	556	2	Q16GN5_AEDAE	Q16gn5 aedes aegypti	RT	cDNA.*;
17	502.5	28.1	556	2	Q01G10_AEDAE	Q01g10 aedes aegypti	RL	Proc. Natl. Acad. Sci. U.S.A. 82:7289-7293(1985).
18	495	27.1	458	2	Q8MQ76_CAEL	Q8mq76 caenorhabditis	RN	[2]
19	495	27.1	560	2	Q29BP0_DROPS	Q29bp0 drosophila	RP	NUCLEOTIDE SEQUENCE [MRNA] (ISOFORM SHORT).
20	453	25.3	551	2	Q8IMY3_DROME	Q8imy3 drosophila	RC	TISSUE=hepatoma;
21	453	25.3	561	2	Q9VCJ4_DROME	Q9vcj4 drosophila	RX	MEDLINE=86085853; PubMed=3001061;
22	453	25.3	573	2	Q4VAF5_DROME	Q4vaf5 drosophila	RA	Tsuji S., Choudhary P.V., Martin B.M., Winfield S., Barranger J.A.,
23	453	25.3	577	2	Q4VAF1_DROME	Q4vaf1 drosophila	RA	Glins E.I.;
24	444	24.8	561	2	Q16581_CAEL	Q16581 caenorhabditis	RT	"Nucleotide sequence of cDNA containing the complete coding sequence
25	441.5	24.7	523	2	Q16580_CAEL	Q16580 caenorhabditis	RT	for human lysosomal glucocerebrosidase.*;
26	441	24.6	524	2	Q611J0_CAERB	Q611j0 caenorhabditis	RL	J. Biol. Chem. 261:50-53(1986).
27	440.5	24.6	495	2	Q611J5_CAERB	Q611j5 caenorhabditis	RN	[3]
28	432.5	24.1	572	2	Q8ATZ7_PHYIN	Q8atz7 phytophthora	RP	NUCLEOTIDE SEQUENCE [GENOMIC DNA].
29	421	23.5	424	2	Q4VAF3_DROME	Q4vaf3 drosophila	RC	TISSUE=liver;
30	409	22.8	423	2	Q7PKY3_ANOGA	Q7pkxy3 anopheles g	RX	MEDLINE=89122038; PubMed=2914709; DOI=10.1016/0898-7543(89)90319-4;
31	392.5	21.9	445	2	A4KMF5_CALSA	A4kmf5 caldicellul	RA	Horowitz M., Wilner S., Horowitz Z., Reiner O., Gelbart T.,
32	376.5	21.0	468	2	Q2AI75_PFTM	Q2ai75 halothermot	RA	Beutler E.;
33	369.5	20.6	495	2	A3J0N5_PFLAO	A3j0n5 flavobacter	RT	"The human glucocerebrosidase gene and pseudogene: structure and
34	369	20.6	443	2	Q8R5Q0_THEIN	Q8r5q0 thermoanaer	RT	evolution.*;
35	345.5	19.3	494	2	A6EQG6_PSPHI	A6eqg6 pedobacter	RL	Genomics 4:87-96(1989).
36	340.5	19.0	445	2	Q1FLM6_CLOST	Q1flm6 clostridium	RN	[4]
37	340.5	19.0	548	2	Q8RPF2_XANCP	Q8rpf2 xanthomonas	RP	NUCLEOTIDE SEQUENCE [GENOMIC DNA].
38	331	18.5	499	2	Q1VR53_PFLAO	Q1vr53 psychroflex	RC	TISSUE=liver;
39	330.5	18.5	474	2	A27WU2_PFLAO	A27wu2 polaribacte	RX	MEDLINE=92241881; PubMed=572652; DOI=10.1016/0898-7543(92)90311-F;
40	328	18.3	489	2	Q3BMD0_XANCS	Q3bmd0 xanthomonas	RA	Beutler E., West C., Gelbart T.;
41	324	18.1	458	2	Q8PM97_XANAC	Q8pm97 xanthomonas	RT	"Polymorphisms in the human glucocerebrosidase gene.*;
42	321.5	18.0	439	2	Q4URV7_XANCS	Q4urv7 xanthomonas	RL	Genomics 12:795-800(1992).
43	318.5	17.8	236	2	Q661W2_XENLA	Q661w2 xenopus lae	RN	[5]
44	318.5	17.8	487	2	A5FJN1_CYTJO	A5fjn1 flavobacter	RP	NUCLEOTIDE SEQUENCE [MRNA] (ISOFORMS LONG AND 3), VARIANTS GO ARG-223;
45	313.5	17.5	490	2	Q1YU93_GAMDM	Q1yu93 gamma prote	RP	GLY-230; PRO-235; ARG-241; ILE-252 AND ARG-364, AND VARIANTS GLY-310

ALIGNMENTS

RESULT 1	
GLUC_HUMAN	
ID	GLUC_HUMAN ← Reviewed; 536 AA.
AC	P04062; Q16545; Q4VXZ2; Q61986; Q9UM03;
DT	01-NOV-1986, integrated into UniProtKB/Swiss-Prot.
DT	09-NOV-2004, sequence version 3.
DT	21-AUG-2007, entry version 105.
DE	Glucosylceramidase precursor (EC 3.2.1.45) (Beta-glucocerebrosidase)
DE	(Acid beta-glucosidase) (D-glucosyl-N-acylsphingosine glucosyltransferase)
DE	(Alglucerase) (Imiglucerase).
GN	Name=GBA; Synonyms=GC, GLUC;
OS	Homo sapiens (Human).
OC	Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi;
OC	Mammalia; Eutheria; Euarchontoglires; Primates; Haplorhina;
OC	Catarrhini; Hominoidea; Homo.
OK	NCBI_TextID=9606;
RN	[1]
RP	NUCLEOTIDE SEQUENCE [MRNA] (ISOFORM SHORT).
RC	TISSUE=placenta;
RX	MEDLINE=86042651; PubMed=3864160;
RA	Sorge J., West C., Westwood B., Beutler E.;
RT	"Molecular cloning and nucleotide sequence of human glucocerebrosidase
RT	cDNA.*;
RL	Proc. Natl. Acad. Sci. U.S.A. 82:7289-7293(1985).
RN	[2]
RP	NUCLEOTIDE SEQUENCE [MRNA] (ISOFORM SHORT).
RC	TISSUE=hepatoma;
RX	MEDLINE=86085853; PubMed=3001061;
RA	Tsuji S., Choudhary P.V., Martin B.M., Winfield S., Barranger J.A.,
RA	Glins E.I.;
RT	"Nucleotide sequence of cDNA containing the complete coding sequence
RT	for human lysosomal glucocerebrosidase.*;
RL	J. Biol. Chem. 261:50-53(1986).
RN	[3]
RP	NUCLEOTIDE SEQUENCE [GENOMIC DNA].
RC	TISSUE=liver;
RX	MEDLINE=89122038; PubMed=2914709; DOI=10.1016/0898-7543(89)90319-4;
RA	Horowitz M., Wilner S., Horowitz Z., Reiner O., Gelbart T.,
RA	Beutler E.;
RT	"The human glucocerebrosidase gene and pseudogene: structure and
RT	evolution.*;
RL	Genomics 4:87-96(1989).
RN	[4]
RP	NUCLEOTIDE SEQUENCE [GENOMIC DNA].
RC	TISSUE=liver;
RX	MEDLINE=92241881; PubMed=572652; DOI=10.1016/0898-7543(92)90311-F;
RA	Beutler E., West C., Gelbart T.;
RT	"Polymorphisms in the human glucocerebrosidase gene.*;
RL	Genomics 12:795-800(1992).
RN	[5]
RP	NUCLEOTIDE SEQUENCE [MRNA] (ISOFORMS LONG AND 3), VARIANTS GO ARG-223;
RP	GLY-230; PRO-235; ARG-241; ILE-252 AND ARG-364, AND VARIANTS GLY-310
RP	AND HIS-369.
RX	MEDLINE=94124033; PubMed=8294033; DOI=10.1016/0378-1119(93)90497-Q;
RA	Inai K., Nakamura M., Yamada M., Asano A., Yokoyama S., Tsuji S.,
RA	Glins E.I.;
RT	"A novel transcript from a pseudogene for human glucocerebrosidase in
RT	non-Gaucher disease cells.*;
RL	Gene 136:365-368(1993).
RN	[6]
RP	NUCLEOTIDE SEQUENCE [GENOMIC DNA].
RX	MEDLINE=97474796; PubMed=9331372;
RA	Winfield S.I., Tayebi N., Martin B.M., Glins E.I., Sidransky E.;
RT	"Identification of three additional genes contiguous to the
RT	glucocerebrosidase locus on chromosome 1q21: implications for Gaucher
RT	disease.*;
RL	Genome Res. 7:1020-1026(1997).
RN	[7]
RP	NUCLEOTIDE SEQUENCE [LARGE SCALE GENOMIC DNA].
RX	PubMed=16710414; DOI=10.1038/nature04727;
RA	Gregory S.G., Barlow K.F., McLeay K.E., Kaul R., Swarbreck D.,
RA	Dunham A., Scott C.E., Howe K.L., Woodfine K., Spencer C.C.A.,
RA	Jones M.C., Gillson C., Searle S., Zhou Y., Kocinski F.,
RA	McDonald L., Evans R., Phillips K., Atkinson A., Cooper R., Jones C.,
RA	Hall R.E., Andrews T.D., Lloyd C., Ainscough R., Almeida J.P.,
RA	Amrose K.D., Anderson P., Andrew R.W., Ashwell R.I.S., Rubin K.,
RA	Babbage A.K., Beggall C.L., Bailey J., Beasley H., Bethal G.,
RA	Bird C.P., Bray-Allen S., Brown J.Y., Brown A.J., Buckley D.,
RA	Burton J., Bye J., Carder C., Chapman J.C., Clark S.Y., Clarke G.,

RA Clee C., Cobley V., Collier R.E., Corby N., Coville G.J., Davies J.,
 RA Deadman R., Dunn M., Bartholomew M., Ellington A.G., Errington R.,
 RA Frankish A., Frankland J., French L., Garner P., Garnett J., Gay L.,
 RA Ghori M.R.J., Gibson R., Gilby L.M., Gillett W., Glithero R.J.,
 RA Graffan D.V., Griffiths C., Griffiths-Jones S., Grocock R.,
 RA Hammond S., Harrison E.S.I., Hart E., Haugen E., Heath P.D.,
 RA Holmes S., Holt K., Howden P.J., Hunt A.R., Hunt S.E., Hunter G.,
 RA Isherwood J., James R., Johnson C., Johnson D., Joy A., Kay M.,
 RA Kershaw J.K., Kikukawa M., Kimberley A.M., King A., Knights A.J.,
 RA Lad H., Laird G., Lawler S., Leongamornlert D.A., Lloyd D.M.,
 RA Loveland J., Lovell J., Lush M.J., Lyne R., Martin S.,
 RA Mashreghi-Mohammadi M., Matthews L., Matthews N.S.W., McLaren S.,
 RA Milne S., Mistry S., Moore M.J.F., Mickerson T., O'Dell C.N.,
 RA Oliver K., Palmeiri A., Palmer S.A., Parker A., Patel D., Pearce A.W.,
 RA Peck A.I., Pelan S., Phelps K., Phillimore B.J., Plumb R., Rajan J.,
 RA Raymond C., Rouse G., Saengphumachak C., Sehra R.K., Sheridan E.,
 RA Showkeen R., Sims S., Skuce C.D., Smith M., Steward C.,
 RA Subramanian S., Sycamore N., Tracey A., Tromans A., Van Helmond L.,
 RA Wall M., Wallis J.M., White S., Whitehead S.L., Wilkinson J.E.,
 RA Willey D.L., Williams H., Wilming L., Wray P.W., Wu Z., Coulson A.,
 RA Vaudin M., Sulston J.E., Durbin R.M., Hubbard T., Wooster R.,
 RA Dunham I., Carter N.F., McVean G., Ross M.T., Harrow J., Olson M.V.,
 RA Beck S., Rogers J., Bentley D.R.;
 RT "The DNA sequence and biological annotation of human chromosome 1.";
 RL Nature 441:315-321 (2006).
 RN [8]
 RP NUCLEOTIDE SEQUENCE [LARGE SCALE MRNA] (ISOPFORM LONG).
 RC TISSUE=Placenta;
 RX PubMed=15489334; DOI=10.1101/gr.2596504;
 RG The MGC Project Team;
 RT "The status, quality, and expansion of the NIH full-length cDNA
 RT project: the Mammalian Gene Collection (MGC).";
 RL Genome Res. 14:2121-2127 (2004).
 RN [9]
 RP NUCLEOTIDE SEQUENCE [GENOMIC DNA / MRNA] OF 1-11.
 RX MEDLINE=68195776; PubMed=3559914;
 RA Reiner O., Wigderson M., Horowitz M.;
 RT "Structural analysis of the human glucocerebrosidase genes.";
 RL DNA 7:107-116 (1988).
 RN [10]
 RP NUCLEOTIDE SEQUENCE [GENOMIC DNA] OF 1-45.
 RX MEDLINE=68074307; PubMed=3687939;
 RA Sorpe J.A., West C., Kuhl W., Treger L., Beutler E.;
 RT "The human glucocerebrosidase gene has two functional ATG initiator
 RT codons.";
 RL Am. J. Hum. Genet. 41:1016-1024 (1987).
 RN [11]
 RP PROTEIN SEQUENCE OF 40-44.
 RC TISSUE=Placenta;
 RA Martin B.M., Murray G.J., Coligan J.E., Raum M., Brady R.O.,
 RA Barranger J.A.;
 RT "Structural studies of human placental glucocerebrosidase.";
 RL Fed. Proc. 43:1869-1869 (1984).
 RN [12]
 RP NUCLEOTIDE SEQUENCE [MRNA] OF 403-416.
 RX MEDLINE=85022513; PubMed=6091633;
 RA Ginn E.L., Choudary P.V., Martin B.M., Winfield S., Stubblefield B.,
 RA Mayor J., Merkle-Lehman D., Murray G.J., Bowers L.A., Barranger J.A.;
 RT "Isolation of cDNA clones for human beta-glucocerebrosidase using the
 RT lambda gT11 expression system.";
 RL Biochem. Biophys. Res. Commun. 123:574-580 (1984).
 RN [13]
 RP NUCLEOTIDE SEQUENCE [GENOMIC DNA] OF 409-462, AND VARIANT GD SER-409.
 RC TISSUE=Skin;
 RX MEDLINE=68176943; PubMed=3533383;
 RA Teuji S., Martin B.M., Barranger J.A., Stubblefield B.K.,
 RA LaMarca M.E., Ginn E.L.;
 RT "Genetic heterogeneity in type 1 Gaucher disease: multiple genotypes
 RT in Ashkenazic and non-Ashkenazic individuals.";
 RL Proc. Natl. Acad. Sci. U.S.A. 85:2349-2352 (1988).
 RN [14]
 RP PROTEIN SEQUENCE OF 469-520.
 RC TISSUE=Placenta;
 RX MEDLINE=60149363; PubMed=3456607;
 RA Dinur T., Oseleki K.M., Legler G., Gatt S., Desnick R.J.,
 RA Grabowski G.A.;
 RT "Human acid beta-glucosidase: isolation and amino acid sequence of a
 RT peptide containing the catalytic site.";
 RL Proc. Natl. Acad. Sci. U.S.A. 83:1660-1664 (1986).
 RN [15]
 RP MASS SPECTROMETRY, MUTAGENESIS OF GLU-379, AND ACTIVE SITE.
 RX PubMed=7908905;
 RA Miao S., McCarter J.D., Grace M.E., Grabowski G.A., Hebersold R.,
 RA Withers S.G.;
 RT "Identification of Glu340 as the active-site nucleophile in human
 RT glucocerebrosidase by use of electrospray tandem mass spectrometry.";
 RL J. Biol. Chem. 269:10975-10978 (1994).
 RN [16]
 RP INTERACTION WITH SAPOSIN-C AND MEMBRANES CONTAINING ANIONIC
 RP PHOSPHOLIPIDS.
 RX PubMed=10781797; DOI=10.1016/S0014-5793(00)01417-4;
 RA Salvio R., Tatti M., Clafford P., Vaccaro A.M.;
 RT "Further studies on the reconstitution of glucosylceramidase activity
 RT by Sap C and anionic phospholipids.";
 RL FEBS Lett. 472:17-21 (2000).
 RN [17]
 RP GLYCOSYLATION AT ASN-98; ASN-185 AND ASN-309.
 RX MEDLINE=22660472; PubMed=12754519; DOI=10.1038/nbt827;
 RA Zhang H., Li X.-J., Martin D.R., Hebersold R.;
 RT "Identification and quantification of N-linked glycoproteins using
 RT hydrazide chemistry, stable isotope labeling and mass spectrometry.";
 RL Nat. Biotechnol. 21:660-666 (2003).
 RN [18]
 RP X-RAY CRYSTALLOGRAPHY (2.0 ANGSTROMS) OF 40-536, GLYCOSYLATION AT
 ASN-58, AND DISULFIDE BONDS.
 RX PubMed=12792654; DOI=10.1038/sj.embo.embor873;
 RA Dvir H., Harel M., McCarthy A.A., Tokar L., Silman I., Futerman A.H.,
 RA Sussman J.L.;
 RT "X-ray structure of human acid-beta-glucosidase, the defective enzyme
 RT in Gaucher disease.";
 RL EMBO Rep. 4:704-709 (2003).
 RN [19]
 RP X-RAY CRYSTALLOGRAPHY (2.4 ANGSTROMS) OF 40-536 IN COMPLEX WITH
 RP SYNTHETIC INHIBITOR, AND ACTIVE SITE.

RX PubMed:15817452; DOI=10.1074/jbc.M502799200;
RA Premkumar L., Sawkar A.R., Boldin-Adamsky S., Tokar L., Silman I.,
RA Kelly J.W., Puterman A.R., Sussman J.L.;
RT "X-ray structure of human acid-beta-glucosidase covalently bound to
RT conduritol-B-epoxide. Implications for Gaucher disease.*";
RL J. Biol. Chem. 260:23815-23819 (2005).
RW [20]
RP X-RAY CRYSTALLOGRAPHY (2.5 ANGSTROMS) OF 40-536, CHARACTERIZATION OF
RP GD VARIANTS SER-55; GLN-87; ASN-118; GLN-153; LEU-161; VAL-162;

Query Match 94.24; Score 1687; DB 1; Length 536;
Best Local Similarity 67.04; Pred. No. 8.6e-137;
Matches 333; Conservative 1; Mismatches 1; Indels 162; Gaps 1;

Qy 1 ARPCIPKSFYSSVWVCNATYCDSEFPTTPALGTFSTRYESTRSGRMELSMGPQANH 60
|||
Db 40 ARPCIPKSFYSSVWVCNATYCDSEFPTTPALGTFSTRYESTRSGRMELSMGPQANH 99

Qy 61 TGTGLLLTLQPEKFKQKVGKGGAGMTDAAALNLALSPPAQULLKSYFSEG----- 113
|||
Db 100 TGTGLLLTLQPEKFKQKVGKGGAGMTDAAALNLALSPPAQULLKSYFSEGIGTHTIR 159

Qy 114 ----- 113

Db 160 VPMASCDPSIRITYADTPDQGLNFSLPERDTKLKPLIHRALQAGRPVSLIASPT 219

Qy 114 ----- 113

Db 220 SPTMLKTNAGVNGKGLKGGPGDIYHQTWARYFYVFLDAYAEHKLQFWNTADNEPSAGL 279

Qy 114 -----VRLIMLNDQRLLPWMAKVVLTQPE 138
|||
Db 280 LSGYFQCLGFTPEKQDFIAROLGPTLANSHTHNWRLMLDQRLLPWMAKVVLTQPE 339

Qy 139 AAKYVHGIANHWYLDLAPAKATLGETHRLFPNTMLFASEACVGSKWQSVLQSSWDG 196
|||
Db 340 AAKYVHGIANHWYLDLAPAKATLGETHRLFPNTMLFASEACVGSKWQSVLQSSWDG 399

Qy 199 MQYSHSIITNLLYHVGMTDNLALPPEGGNWNVNFVDSPIVDITKOTFYQPMFYEL 258
|||
Db 400 MQYSHSIITNLLYHVGMTDNLALPPEGGNWNVNFVDSPIVDITKOTFYQPMFYEL 459

Qy 259 GHFSKFIPEGSRWGLVASQRNDLDAVALMHPDGSANVWVLRSSKNVPLTIKDPANGVL 318
|||
Db 460 GHFSKFIPEGSRWGLVASQRNDLDAVALMHPDGSANVWVLRSSKNVPLTIKDPANGVL 519

Qy 319 ETISPGYSIHTYLARRQ 335
|||
Db 520 ETISPGYSIHTYLARRQ 536

RESULT 2
GLCH_PANTR
ID GLCH_PANTR Reviewed; 536 AA.
AC Q3HOT0;
DT 21-NOV-2003, integrated into UniProtKB/Swiss-Prot.
DC 01-JUN-2001, sequence version 1.